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An Algorithm Approach to Pediatric Diagnosis

Approach to Short Stature

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Definition

- Height 2 standard deviation score (SDS) below the mean for age and sex, height <2 SDS of the target height (TH)
- A significant decrease in height SDS defined as a deflection of at least 0.3 SDS/yr

Etiology

Causes of short stature:

- *Isolated short stature:*
  - Idiopathic short stature (ISS)
  - Familial short stature (FSS)
  - Constitutional delay in growth and puberty (CDGP)
- *Syndromic short stature:*
  - Turner syndrome
  - Noonan syndrome
  - Silver–Russel syndrome
  - Prader–Willi syndrome

- *Disorders of the growth hormone (GH)–insulin-like growth factor (IGF)-I axis:*
  - Growth hormone deficiency (GHD)
  - Growth hormone insensitivity
- *Chronic systemic diseases:*
  - Celiac disease
  - Chronic kidney disease
  - Cardiac conditions
  - Pulmonary diseases
  - Neuromuscular disorders
  - Endocrine conditions
  - Gastrointestinal disorders
  - Protein energy malnutrition
  - Rheumatic disorders
  - Hematological disorders
- *Skeletal dysplasia:*
  - Achondroplasia
  - Hypochondroplasia
- *Small for gestational age babies (SGA)*
- *Psychosocial deprivation*

Red Flag Signs

- Height < –3 SD for age and sex
- Growth velocity <25th percentile
- Abnormal body proportions
- Dysmorphic features
- Abnormal central nervous system (CNS)/eye findings
- Goiter

Clinical Evaluation

Anthropometry

Anthropometry—weight, standing height (stadiometer)/length by infantometer (<2 years), proportion (upper segment: lower segment), head circumference, arm span.

Growth Chart

It is recommended that the measurements should be plotted and evaluated using country specific charts, that is combined World Health Organization/Indian Academy of Pediatrics (WHO/IAP) growth charts.

Growth Velocity

A series of height measurements indicates the growth pace. Poor prognosis for height if below the 25th centile.

T1: Height measured previously. T2: Height measured thereafter

$$\text{Growth velocity (cm/yr)} = \frac{\text{T2} - \text{T1}}{\text{Number of months between T2 and T1}} \times 12 \text{ months}$$

Growth Potential

The midparental height (MPH) is used in order to identify FSS. The MPH for boys and girls is determined differently, as follows:

- MPH (Boys) = (Father's height + Mother's height + 13)/2
- MPH (girls) = (Father's height + Mother's height – 13)/2

Midparental height gives us an idea about the growth potential genetically endowed on the child.

Pointers to etiology in clinical evaluation are given in **Table 1.**

TABLE 1: Pointers to etiology in clinical evaluation.

Pointers	*Etiology*
Skull	Large anterior fontanelle—achondroplasia, MPS
Facies	Cherubic facies—GHD, Gargoyle facies—MPS
Eyes	Cataract—intrauterine infections, corneal clouding—MPS
Teeth	Delayed dentition—hypothyroidism, GHD, rickets
Neck, chest	Webbed neck, shield chest—Turner syndrome
Limbs	Deformities in skeletal dysplasia, rickets
Genitalia	Cryptorchidism in hypopituitarism

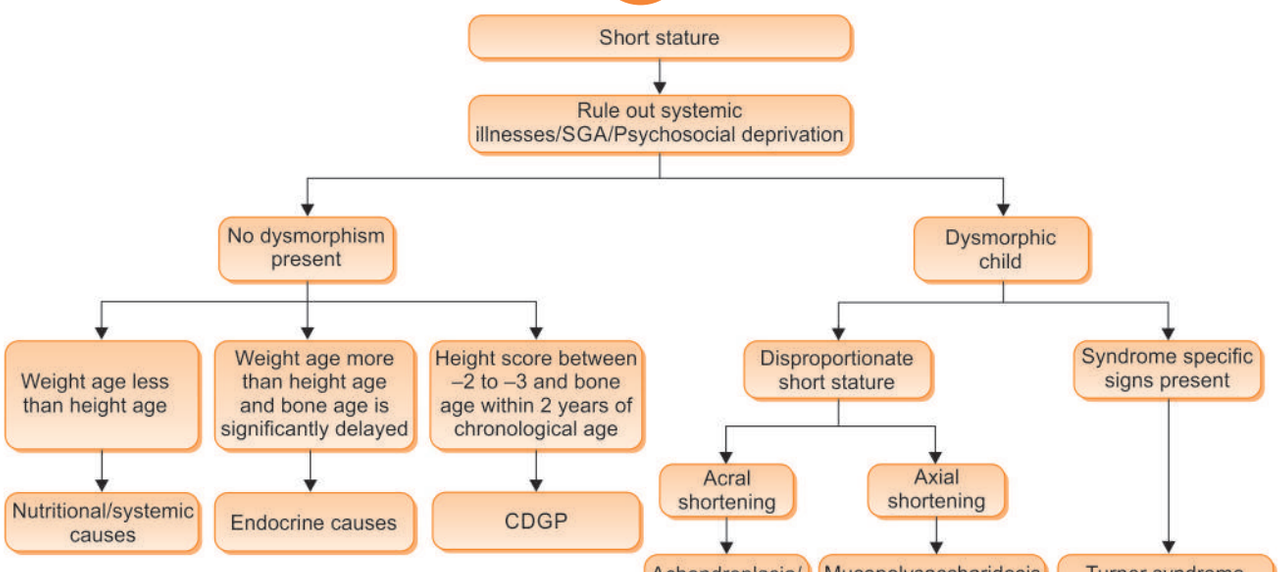
(GHD: growth hormone deficiency; MPS: mucopolysaccharidoses)

Investigations

FIRST-LINE	SECOND-LINE
CBC, ESR, CRP	Growth hormone stimulation test—at least two growth provocation tests done in suspected isolated GHD (stimulating agent—clonidine/arginine/insulin/glucagon/dopamine/GHRH)
Renal and liver function test	MRI brain
Venous blood gas, electrolytes	Genetic test
Calcium, phosphorus, alkaline phosphatase	
Thyroid function test	
Celiac screen	
• Insulin-like growth factor 1 (IGF1) • IGF-binding protein 3 (IGFBP3) in children <2 years	
Bone age	
Karyotyping in girls with unexplained short stature	

(CBC: complete blood count; CRP: C-reactive protein; ESR: erythrocyte sedimentation rate; GHD: growth hormone deficiency; GHRH: growth hormone-releasing hormone)

Approach to Short Stature



(SGA: small for gestational age; CDGP: constitutional delay in growth and puberty)

Suggested Reading

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